A novel homozygous frameshift *SPTBN2* gene mutation associated with Spinocerebellar Ataxia-14

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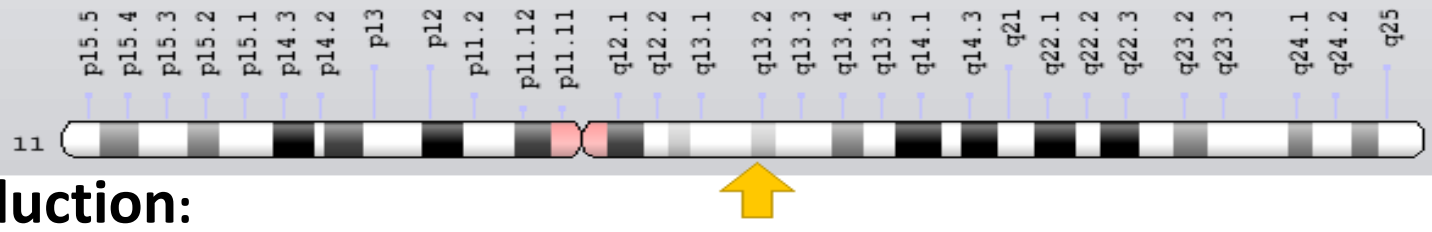
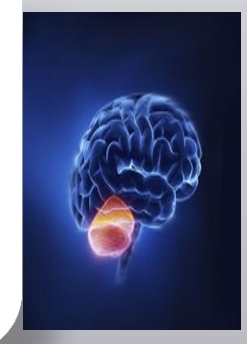
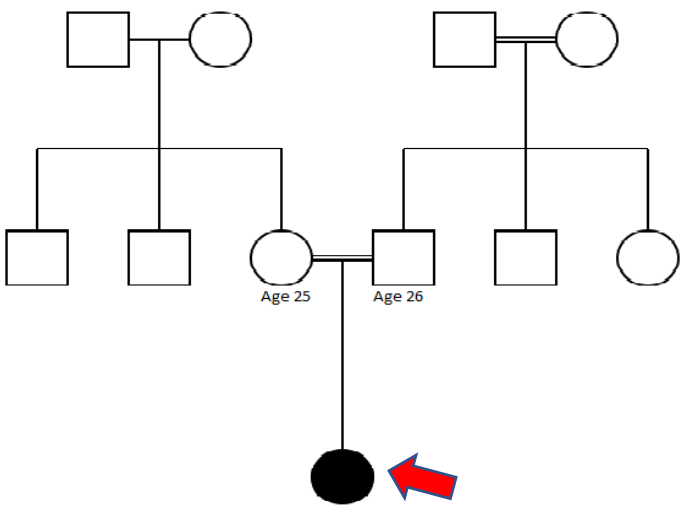


Introduction:

Spinocerebellar ataxia-14 caused by a homozygous mutation in the *SPTBN2* gene, is a neurologic disorder characterized by delayed psychomotor development, severe early-onset gait ataxia, eye movement abnormalities, cerebellar atrophy on brain imaging, and intellectual disability. Unlike many other spinoserebellar ataxia, it is infantile onset.



Pedigri Chart:



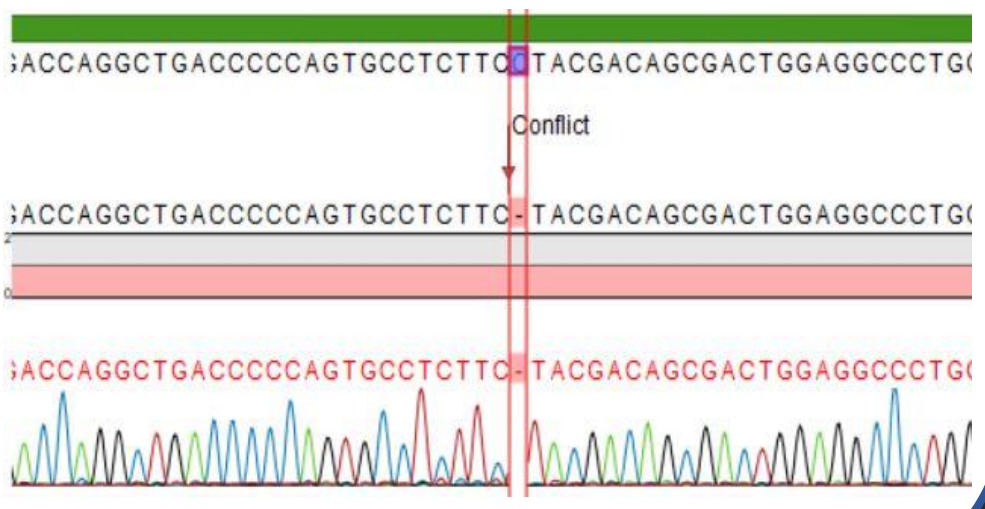
Methodology:

- DNA extraction from patient's whole blood
- Library preparation with 50 ng genomic DNA using Illumina Nextera Exom Sequencing Kit
- Sequencing with Illumina Nextseq 500 platform
- Analysis data and annotate variants
- Sanger Sequencing for Confirmation

Results:

- ✓ We detected a homozygous frameshift deletion, c.3427delC, also confirmed via Sanger sequencing, on *SPTBN2* gene. This variant was also observed heterozygous state in her mother and father who had relationship between them.
- ✓ As far as we know, this frameshift deletion is not found any clinical databases (ClinVar or Human Genome Database (HGMD) ect.). It could alter gene function by causing an early termination of gene expression. According to the guideline of American College of Medical Genetics (ACMG) it is classified as 'pathogenic.'

Integrative Genomic Viewer (IGV) Image



Mutation Detection by Sanger Sequencing

Conclusion:

As a conclusion, with this mutation that we found , we added a new one to the variants detected in the *SPTBN2* gene. Functional studies are needed to contribute to the determination of the pathogenicity of this variant.

Acknowledgements:

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